MOLECULAR DIAGNOSTICS UNIT

Luis Lombardía
UNI Head

Technician
Diana Romero

OVERVIEW

The Molecular Diagnostics Unit’s (MDU) commitment to quality molecular testing, in both clinical and laboratory settings, ensures comprehensive support for cancer patient care and cancer research efforts by aligning with the growing needs of healthcare professionals and researchers. Thus, MDU plays a role in the Spanish healthcare system by offering a range of molecular diagnostic tests that aid clinicians in early cancer diagnosis, the detection of relapses, and monitoring therapy responses. These assays are constantly revised, integrating the latest diagnostic tests and upgrading the established ones. Likewise, the Unit provides support to CNIO’s Research Groups by analysing their samples for specific biomarker alterations or by providing specialised technical assistance. MDU is at the forefront of molecular diagnostics standardisation, collaborating with international and national organisations. Finally, through laboratory training, the Unit coaches biomedical students, technicians, and residents.

“IVDR implementation will improve the accuracy, reliability and efficiency of cancer diagnostics testing, thus ensuring improved patient care.” (FIGURE 1)

During 2023, our catalogue grew with the addition of a new assay. This assay will enable the detection, through direct sequencing, of p.C515S substitution in exon 15 of the BTK (Bruton tyrosine kinase) gene that mediates resistance to a BTK inhibitor, ibrutinib, by affecting its covalent binding to BTK. As a result, the detection of this mutation will help haematologists to switch the treatment of patients with chronic lymphocytic leukaemia by using second line non-covalent BTK inhibitors.

We have also improved the clinical utility of 2 assays listed in our catalogue. Firstly, to amend the diagnosis, prognosis and/or personalised therapy of cancer patients, especially those with acute myeloid leukaemia, we have extended the mutation detection scope in the TP53 gene - previously limited to exons 5, 6, 7 and 8 - to its whole sequence. Likewise, the detection of recurrent mutations already done in exons 9, 11, 13 and 17 of the KIT gene has been extended to exon 18. This upgrade will allow oncologists to offer new therapeutic options to their patients with gynaecological melanomas that harbour mutations in the KIT gene, not previously detected in cutaneous melanomas.

Additionally, last summer, aiming to replace the existing European In Vitro Diagnostic Directive (IVDD), we launched a key development that will lead us to implement a new In Vitro Diagnostic Regulation (IVDR) that is mandatory for all CNIO’s diagnostics support units in the midterm (FIGURE 1). To comply with IVDR requirements, our efforts address the upgrading and validation of the current assays to guarantee their safety and their analytical and clinical performance. With the aid of experts in Quality Management Systems, we are already beginning to establish robust quality control and assurance procedures, as well as documentation schemes that will warrant firm compliance with IVDR guidelines.

Finally, during 2023, in the framework of our training policy, we hosted an undergraduate student in biomedical engineering.

CORE UNIT HIGHLIGHTS

“IVDR implementation will improve the accuracy, reliability and efficiency of cancer diagnostics testing, thus ensuring improved patient care.” (FIGURE 1)